

Prenatal diagnosis and postnatal findings of cloacal malformation: a case report

Case Report

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Abstract:

Introduction: Cloacal malformation is an extremely rare fetal pathological condition but an important anomaly that presents as a variety of defects. It predominantly affects females, with prevalence of 1 in 50,000 births.

Prenatal ultrasonography usually showed oligohydramnios and the fetus having a large cystic mass in the lower abdomen with a single septum, bilateral hydronephrosis, ambiguous genitalia and a single umbilical artery. Postnatal finding including ambiguous genitalia and rectal atresia with a single perineal opening.

Case report: The purpose of this case series was to illustrate characteristic prenatal sonographic features of cloacal malformation by using imaging from 3 cases seen in all academic centers to augment published data. The imaging feature common to all cases was a central cystic pelvic mass containing a characteristic fluid-fluid level.

Conclusions: The children were followed at regular intervals by an urologist, surgeon and nephrologist throughout life. Medical teams were committed to providing long-term care for children with cloacal anomalies.

Keywords: Cloacal Malformation, Defects, Prenatal Sonographic

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Introduction:

Embryogenesis of the cloacal sinus, anorectal canal and lower urinary tract includes the development of the mesonephric duct and urogenital sinus. The mesonephric duct from which the ureteric buds arise inserts into the lower allantois, just above the terminal part of the hindgut, the cloaca. After that, mesoderm proliferates and forms the transverse mesodermal ridge, the urorectal septum that divides the cloaca into the anterior portion, the primitive urogenital sinus and the posterior portion, the cloacal sinus or anorectal canal. The mesonephric ducts open into the urogenital sinus and later become the ureters. The urorectal septum develops caudally and fuses with the cloacal membrane, dividing it into the urogenital membrane (anterior) and the anorectal membrane (posterior) by the end of the seventh week. The primitive perineal body forms at the site of fusion ^[1, 2]. The cloacal malformation is an extremely rare fetal pathological condition but an important anomaly that presents as a variety of defects. It predominantly affects females, with prevalence of 1 in 50,000 births. The cloacal malformation or anomaly is a complex congenital problem defined by a confluence or coming together of the urinary tract, vagina and rectum to form a single common channel that opens on the perineum. It is common channel that is referred to as a cloaca. The discovery of a megacyst during the first trimester may indicate a diagnosis of cloacal malformation ^[3].

Relatively in 75% of cases anorectal, canal malformation is diagnosed as part of VACTERL/ VATER syndrome (include the birth defects of vertebral anomalies, Anal Atresia, Cardiac defects, Tracheo-esophageal fistula and/or Esophageal atresia, Renal anomalies and Limb defects) [4,5]. Occasionally, this diagnosis may be considered when a combination of abnormalities of the bladder and vagina are detected antenatally, but most often, it is diagnosed during the immediate newborn period due to discovery of a single perineal opening. The cause of this defect is unknown [6]. Initially, the goals are to identify the associated abnormalities, to provide drainage of the urinary tract and to divert the bowel by using a colostomy. While the initial goal is to stabilize the child and relieve blockages in the urinary and intestinal tract, the long-range goals are directed at restoring anatomy and function. [7, 8, 9]. Prenatal diagnosis is possible with knowledge of the distinctive imaging features. Antenatal ultrasound for determine the prognosis for women presenting with preterm labor and intact membranes. Antenatal ultrasound and Magnetic Resonance Imaging (MRI) have been used to detect the full range of fatal malformations [10].

Cloacal-related anomalies can also result in multiple vaginas, a malformed anus and other defects of the ureters and kidneys. A commonly associated genitourinary abnormality occurring in 50% of cases is hydrometrocolpos which is a fluid collection within the bladder and uterus that may press on the base of the bladder neck leading to obstruction of the bladder. Duplication of the renal collecting system is the commonest major congenital malformation of the urinary tract. To assess the accuracy of ultrasound diagnosis of structural anomaly kidneys in the fetus and to identify prognostic features of antenatal sonography associated with this diagnosis. When such problems are detected neurosurgeons, neurologists, pediatric nephrologists and orthopedists are consulted [11,12].

The children are followed by an urologist and nephrologist in their life. Medical teams are committed to providing long-term care for children with cloacal anomalies. Generally, patients are able to live happy, energetic productive lives with bowel and bladder social continence. The purpose of this case series was to illustrate the characteristic prenatal sonographic features of cloacal malformation by using imaging from 3 cases seen in all academic centers to augment published data.

Case presentation:

Case 1: Prenatal ultrasonography on a 31-year-old woman at 33 weeks of pregnancy showed the fetus having a large cystic mass in the lower abdomen with a single septum, bilateral hydronephrosis, ambiguous genitalia and a single umbilical artery. The pregnancy developed accentuated oligohydr-amnios and the presence of a fetal brain-sparing effect was diagnosed by using arterial Doppler velocimetry. The newborn showed abdominal distension, ambiguous genitalia and rectal atresia with a single perineal opening. Pelvic ultrasound done on her in the first day after the delivery and it was revealed that there was a large retrovesical septated cystic mass of dense content in the fetal abdomen and bilateral hydronephrosis. Hysterotomy was performed and 70 mL of dense liquid was drained through an abdominal colostomy. The infant died on the 28th day of life due to the infectious complications. Additional anomalies included uterine and vaginal duplication, hydronephrosis, and lumbo-sacral anomalies.

Case 2: Fetal female urogenital anomalies are often difficult to evaluate by ultrasonography, especially in late gestation. The researchers had a case of fetal hydrometrocolpos which was detected at 36 weeks of gestation. Ultrasonography revealed a large retrovesical septate hypoechogenic mass in the fetal abdomen; however, the sonographic findings were inconclusive. Ultrasonography confirmed that the abdominal mass was fluid-filled with a mid-plane septum in the midline posterior to the bladder and showed a connection to the dilated uterus that was duplicated. These findings were consistent with a diagnosis of hydrometrocolpos with septate vagina and uterus didelphys. The neonate showed abdominal distension, ambiguous genitalia and anal atresia with a single perineal opening. hydro-metrocolpos was secondary to a urethral type of cloacal anomaly. Aspiration of the mass and a colostomy were performed on the first postnatal day and it was followed by anorectoplasty at 19 months of age. MRI is a useful complementary tool for assessing fetal urogenital anomalies when ultrasonography is inconclusive .

Case 3: Prenatal ultrasonography on a 25-year-old woman at 31 weeks of pregnancy showed the fetus having a large cystic mass in the lower abdomen with a single septum, bilateral hydronephrosis, ambiguous genitalia and a single umbilical artery. The pregnancy developed accentuated oligohydramnios. The newborn showed abdominal distension, ambiguous genitalia and rectal atresia, with a single perineal opening. Pelvic

ultrasound was done on the baby in the first day after delivery and it revealed the presence of a large retrovesical septated cystic mass of dense content in the fetal abdomen and bilateral hydronephrosis. Presence of a septated cyst with dense content in the fetal abdomen confirmed the finding of hydrometrocolpos, thus it was raising the clinical suspicion of a cloacal anomaly.

Discussion:

The management of cloacal malformations requires a collaborative effort by experienced pediatric surgeons, pediatric nephrologist and nephrologists. Initial goal is prenatal diagnosis to identify the associated abnormalities and stabilize the child and to relieve the blockages in the urinary and intestinal tract. The long-range goals are directed at restoring the anatomy and function. Last goal is to provide the drainage of the urinary tract and to divert the bowel by using a colostomy [7]. Antibiotic prophylaxis (low dose once a day antibiotic) was given to prevent urinary tract infections. Imaging studies that were important in the newborn period include ultrasonography of the kidneys, bladder and other pelvic organs (renal bladder, pelvic ultrasound), plain films of the spine and sacrum, spine ultrasound and magnetic resonance imaging.

Often there is dilation of the kidney drainage system (hydronephrosis) and vesicoureteral reflux (regurgitation of urine from the bladder to the kidneys) may be present. Spine and sacral are abnormalities which are common and include hemivertebra (incomplete development of one side of a vertebra) and incomplete development of the sacrum. The purpose of spinal ultrasound or spine MRI is to detect a "tethered cord". This problem exists in 40% of children with cloacal malformations referred to an abnormal position of the spinal cord within the bony spinal column. Great variation exists in anatomy and corrective efforts must be individualized. Further imaging and endoscopy (using a lighted telescope to view internal structures) are necessary before the repairs are performed in order to define the type of repair that is appropriate. Anorectovaginourethoplasty is the name given to simultaneous and comprehensive repair of urethra, vagina and rectum bringing each to a separate opening on the perineum [9].

Even with successful repair, some children may require bladder catheterization to empty their bladders and may remain vulnerable to urinary tract infections. For this reason, children are followed at regular intervals by an urologist in their all life. Cloacal

anomalies require surgical repair. The procedure depends on the type and extent of the abnormality [8].

Anorectal Malformation (ARM) is a relatively common malformation, which can be diagnosed during prenatal ultrasound examination and almost at the end of the first trimester. In the majority of cases, ARM are associated with other malformations, like VACTERL syndrome. Anecho/hypoechoic dilatation of the intestine is the main sonographic feature of this malformation. The children are followed at regular intervals by an urologist, surgeon and nephrologist in their throughout life. Medical teams are committed to provide long-term care for children with cloacal anomalies. Generally, patients are able to live happy, energetic productive lives with bowel and bladder social continence.

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